2022 IMPACT REPORT

Promoting Patient-Focused Drug Development

- Externally-Led Patient-Focused Drug Development
- EL-PFDD meeting for PMS before the FDA
  - held in November 2022
  - nearly 300 families participated
  - more than 150 written comments
  - attended by 13 Companies, 30 Researchers & 16 FDA Regulators
  - $80,000

Phelan-McDermid syndrome CONCEPTUAL MODEL

- data extracted from 15 peer-reviewed publications
- 96 disease signs and symptoms
- represented 13 observer-reportable patient impacts
- 31 caregiver impacts
  - $25,000

Created two new REAL FAMILY STORIES videos

- self-injurious behaviors
- severe hypotonia
  - $10,000

Accelerating Translation of Research to Therapeutics

CureSHANK Biomarkers and Outcome Measures Consortium

- $100,000

- Retained a project manager and legal team
- Engaged with 22 potential industry partners
- Developed governance and terms of participation

PMS Community Engagement

Two Community Webinars

- updating & engaging the PMS community on CureSHANK’s work
- $100,000